

The present invention provides isolated nucleic acids encoding human SCA2 protein, or fragments thereof, and isolated SCA2 proteins encoded thereby. Further provided are vectors containing invention nucleic acids, probes that hybridize thereto, host cells transformed therewith, antisense oligonucleotides thereto and compositions containing antibodies that specifically bind to invention polypeptides, as well as transgenic non-human mammals that express the invention protein. In addition, methods for diagnosing spinocerebellar Ataxia Type 2 are provided.